10q26 deletion syndrome

Description

10q26 deletion syndrome is a condition that results from the loss (deletion) of a small piece of chromosome 10 in each cell. The deletion occurs on the long (q) arm of the chromosome at a position designated 10q26.

The signs and symptoms of 10q26 deletion syndrome vary widely, even among affected members of the same family. Among the more common features associated with this chromosomal change are distinctive facial features, mild to moderate intellectual disability, growth problems, and developmental delay. People with 10q26 deletion syndrome often have delayed development of speech and of motor skills such as sitting, crawling, and walking. Some have limited speech throughout life. Affected individuals may experience seizures, attention-deficit/hyperactivity disorder (ADHD), poor impulse control (impulsivity), or exhibit autistic behaviors that affect communication and social interaction.

A range of facial features is seen in people with 10q26 deletion syndrome, but not all affected individuals have these features. Facial features of people with 10q26 deletion syndrome may include a prominent or beaked nose, a broad nasal bridge, a small jaw (micrognathia), malformed ears that are low set, a thin upper lip, and an unusually small head size (microcephaly). Many affected individuals have widely spaced eyes (hypertelorism) that do not look in the same direction (strabismus). Some people with this condition have a short neck with extra folds of skin (webbed neck).

Less common signs and symptoms can occur in 10q26 deletion syndrome. Skeletal problems include a spine that curves to the side (scoliosis), limited movement in the elbows or other joints, or curved fifth fingers and toes (clinodactyly). Slow growth before and after birth can also occur in affected individuals. Males with this condition may have genital abnormalities, such as a small penis (micropenis), undescended testes (cryptorchidism), or the urethra opening on the underside of the penis (hypospadias). Some people with 10q26 deletion syndrome have kidney abnormalities, heart defects, breathing problems, recurrent infections, or hearing or vision problems.

Frequency

10q26 deletion syndrome is thought to be a rare condition; at least 100 cases have been described in the scientific literature.

Causes

People with a 10q26 deletion syndrome are missing between 3.5 million and 17 million DNA building blocks (base pairs), also written as 3.5-17 megabases (Mb), at position q26 on chromosome 10. The exact size of the deletion varies, and it is unclear what exact region needs to be deleted to cause the condition. In many affected individuals, the 10q26 deletions include the tip of the q arm of chromosome 10; however, some smaller deletions occur within the arm of the chromosome.

The signs and symptoms of 10q26 deletion syndrome are probably related to the loss of one or more genes in the deleted region. However, it is unclear which missing genes contribute to the specific features of the disorder.

Learn more about the chromosome associated with 10q26 deletion syndrome

- chromosome 10

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered chromosome in each cell is sufficient to cause the disorder.

This condition may be inherited or occur as a result of a new chromosomal change. In some cases, an affected person inherits the chromosome with a deleted segment from a parent. Other cases result from a deletion that occurs during the formation of reproductive cells (eggs or sperm) in an affected individual's parent or in early fetal development. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- 10qter deletion
- Chromosome 10q26 deletion syndrome
- Distal 10q deletion syndrome
- Distal deletion 10q
- Distal monosomy 10q
- Monosomy 10qter
- Telomeric deletion 10
- Terminal chromosome 10q26 deletion syndrome

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 10q26 deletion syndrome (https://www.ncbi

.nlm.nih.gov/gtr/conditions/C2674937/)

Patient Support and Advocacy Resources

- Disease InfoSearch (https://www.diseaseinfosearch.org/)
- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- CHROMOSOME 10q26 DELETION SYNDROME (https://omim.org/entry/609625)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%2810q26%5BTI%5D%29+AND+%28deletion%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

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